

Mid-Cavity Obstruction and High Intraventricular Pressure in Child with Critical Hypertrophic Cardiomyopathy



Healthcare

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Abstract

Hypertrophic cardiomyopathy (HCM) is very common and can affect people of any age. About one out of every 500 people has HCM. It affects men and women equally. HCM is a common cause of sudden cardiac arrest (SCA) in young people, including young athletes. HCM occurs if heart muscle cells enlarge and cause the walls of the ventricles (usually the left ventricle) to thicken. Despite this thickening, the ventricle size often remains normal. However, the thickening may block blood flow out of the ventricle. If this happens, the condition is called obstructive hypertrophic cardiomyopathy. HCM also can affect the heart's mitral valve, causing blood to leak backward through the valve. Sometimes, the thickened heart muscle doesn't block blood flow out of the left ventricle. This is called non-obstructive hypertrophic cardiomyopathy. On this presentation we report a case of a child, with a rare form of the idiopathic hypertrophic cardiomyopathy, associated with mid-cavity obstruction and high intraventricular peak pressure. Cardiomyopathy, diagnosed antenatally, was followed postnatally and, despite of a lot echocardiographic findings - the growing, development and clinical signs are minimal.

The most characteristic morphological abnormality in children with hypertrophic cardiomyopathy (HCM) is the excessive hypertrophy of and no dilated left ventricle with absence of other cardiac or systemic diseases that could produce left ventricular hypertrophy [1], [3].

Often, it is asymmetric in nature, with a preference for ventricular septum and occurs either in sporadic or familial forms. From the genetics point of view the disease is highly variable with respect to the specific gene mutation and degree of penetration. Basically, there is no correlation between the severity of the disease and symptomatology. Often, children lack symptoms for a long period of time and frequently, the disease is detected in case of presence murmur or arrhythmia. Classically, symptoms include pulmonary congestion, fatigue, palpitations, chest pain, syncope and congestive heart failure. A number of pathophysiological components and process are identified: systolic dysfunction and left ventricular outflow tract obstruction, diastolic dysfunction, coronary artery abnormalities, leading to myocardial ischemia, mitral regurgitation, arrhythmias and sudden cardiac death [2],[3].

M-mode and 2-D echocardiography are the primary screening and evaluation of HCM tools, whereas Doppler imaging can fully delineate the entire spectrum of hemodynamic abnormalities. During the routine echocardiography screening by an obstetrician hypertrophy of the left ventricle was registered, otherwise, until then, a normal pregnancy of 36 weeks. Left ventricular hypertrophy was present at the second level of fetal echocardiographic examination. Especially hypertrophic was presented interventricular septum, with separate cavity of left ventricle in two parts. Apical part has been seen as separated from the outlet and inlet part, acquiring a diverticular shape.

This part was clearly presented that actively take part in left ventricle contractility. By continuous Doppler waves between the two parts high velocity was measured consistent with the maximum hemodynamic gradient of 37 mmHg at a mid left ventricle level. The Color flow Doppler imaging showed a narrow area of the left ventricular cavity in systole and a trivial mitral regurgitation was noted. This raised doubts that it is more

likely that this is a case of suspected hypertrophic cardiomyopathy then a case of diverticulum of the left ventricle.

Pregnancy went to full term, with a vaginal delivery of a male infant, weighing 3280g, Apgar score was 8/9. Complete clinical and cardiological examination was obtained. An arterial blood gas showed normal range. Clinical examination presented a quite precordium, normal first heart sound, very short midsystolic murmur on the apex and single second heart sound. Electrocardiogram showed non-specific changes, presenting normal sinus rhythm, normal frequency and left heart deviation with signs of biventricular hypertrophy.

A chest radiogram revealed an enlargement cardiac silhouette with a narrow mediastinum. The pulmonary vasculature was normal in appearance, and there were no infiltrates seen. Echocardiography demonstrated a normal systemic and pulmonary vein connection, with small interatrial communication. There was normal atrio-ventricular and ventriculo-arterial connection.

Cross-sectional echocardiography from apical four-chamber view, during the diastola presented hypertrophy of the left ventricle walls, especially interventricular septum. During the systola left ventricular walls form obstruction at the middle level of the left ventricle, causing two “separate chambers” where apical part actively participates during the contraction. The Color flow Doppler imaging shows a very narrow and constricted area at the middle ventricle level in systole whereas continuous Doppler imaging presented a high systolic velocity with the peak gradient of 53 mmHg. Other echocardiography findings, including aortic valve, aortic arch and coronary artery, were normal. Blood pressure was within normal limits. The child’s growth and development was completely normal, there was no sweating or fatigue during physical activities. Last examination was done at the age of 11 months when the child weighted 9450g (50%), and clinically manifested only syderopenic anemia. Final echocardiographic examination registered increasing mid-cavity obstruction at the papillary muscles level and increasing intracavitary pressure, with peak gradient of 105.1 mmHg. In good basic condition child is continually under Propranolol therapy, 1 mg/kg divided in three doses.

Discussion

An atypical form of hypertrophic cardiomyopathy is presented with mid-cavitary obstruction of left ventricle and fast increasing intracavitary pressure. This form involves selective hypertrophy and obstruction at the mid-left ventricular level. In adults, these findings may represent effects of a long-standing hypertension with relatively small left ventricular cavities in some individuals. [2],[8].

In children this type of hypertrophic cardiomyopathy is very rare and it is quite likely that there is a distinct anatomic subtype of hypertrophic cardiomyopathy, with still clearly unidentified etiology. Over the recent years, knowledge about HCM has evolved enormously, mainly through advances in molecular genetics and in understanding of pathophysiological mechanisms, as well as our awareness of the great variability in its expression. HCM is now recognized as a genetic cardiac disease with an autosomal dominant pattern of inheritance, but with variable penetrance and expression albeit that sporadic case occurs[7].

There is genetic heterogeneity, with more than one gene being associated with the clinical manifestation and there is a great phenotypic variability, not only among unrelated families, but also within the same family [1].

All forms of HCM have in common an inappropriate left ventricular hypertrophy, often with abnormal myofibril orientation and represent a diverse spectrum of disease with varying degrees of hypertrophic expression even among a given family. There is, however, wide variation of expression within affected families. It is believed that the penetrance of the disease is incomplete during childhood and adolescence, increasing with age to nearly complete penetrance in adulthood. In general, there is no correlation between the severity of the disease and the symptomatology [3],[6].

Children often lack symptoms, and as a rule, HCM does not interfere with the physical development of the patient except in its severe forms. Affected children may remain asymptomatic for a long period of time. The clinical manifestation of the classic form of HCM result from systolic dysfunction and dynamic left ventricular outflow tract obstruction, diastolic dysfunction, mitral regurgitation, a high prevalence of arrhythmias and sudden cardiac death [4].

Our case is specific in many aspects including early antenatal diagnosis with normal growth without other manifestations during pregnancy, normal at term delivery and asymptomatic neonatal and postnatal period. Our case is especially interesting because of the increasing intracavitary pressure for a short time - during delivery it was 37 mmHg and at the 11th month of life the peak pressure was 105.1 mmHg.

When HCM is suspected, the antenatal detection is relatively easy after elimination of other cardiac diseases caused by the left ventricle hypertrophy (coronary anomalies, congenital heart obstruction like aortic valvular stenosis, aortic coarctation etc) [5].

According to the literature, it is still a small number of children antenatally diagnosed with mid-cavity HCM and possibility of miss diagnosis is high. Also, our case is a rare one because of the existing such high peak intracavitary pressure without any clinical manifestations.

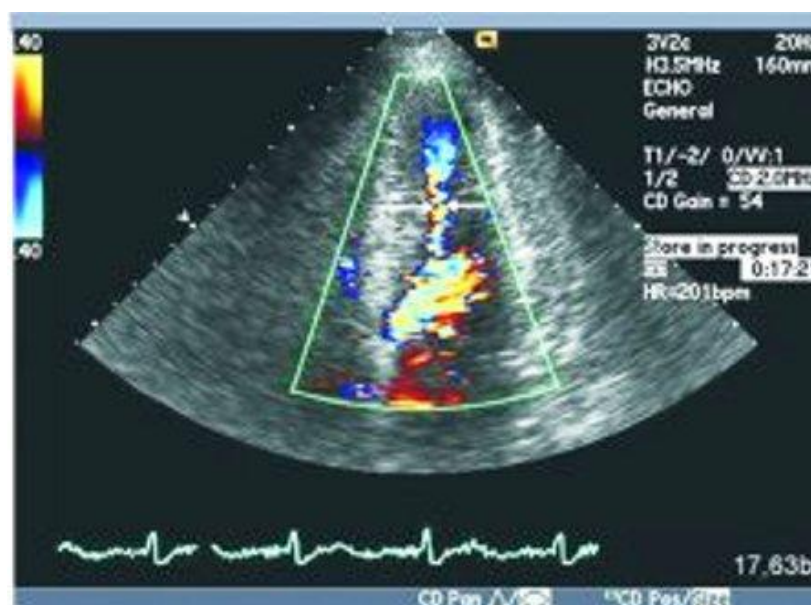


Fig 1. Turbulent flow presented by color Doppler image from apical view of a intracavitary mid-level obstruction of left ventricle

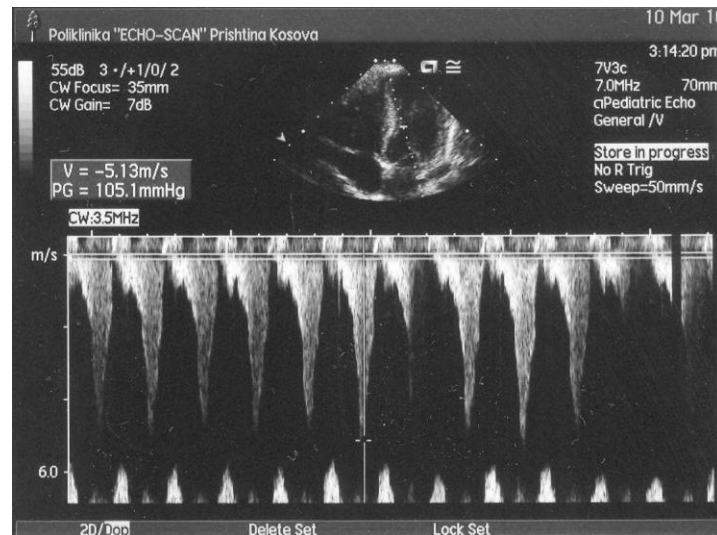


Fig 2. High peak pressure measured by continuous Doppler from apical view

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